10th Science Lesson 18 Notes in English

18] Genetics

Introduction

- "Like Begets Like" is an important and universal phenomenon of life as the living beings produce offspring of their own kind.
- Color of eye, color of hair, shape of nose, type of earlobe, etc, are inheritable traits.
- Some of our characteristics might have been inherited from our grandparents.
- It is because of the genes we inherit from our parents. These genes are responsible for the physical outlook and biological functions.
- The branch of biology that deals with the genes, genetic variation and heredity of living organisms is called genetics.
- Heredity is transmission of characters, from one generation to the next generation, while variation refers to the differences shown by the individuals of the same species and also by the offspring of the same parents.
- All these can happen only due to chromosomes. Now let's see what chromosomes are and how they are composed with DNA that forms the genetic material.

Gregor Johann Mendel – Father of Genetics

- **Mendel** (1822-1884) was an Austrian monk who discovered the basic principles of heredity through his experiments. His experiments are the foundation for modern genetics.
- He was born in 1822 to a family of farmers in Silesian of Czechoslovakia. After finishing his high school at the age of 18, he entered the Augustinian Monastery at Brunn as a priest.
- From there he went to the University of Vienna for training in physics, mathematics and natural science.
- Mendel returned to the monastery in 1854 and continued to work as a priest and teach in high school.
- In his leisure time he started his famous experiments on the garden pea plant.
- He conducted his experiments in the monastery for about nine years from 1856 to 1865. He had worked on nearly 10000 pea plants of 34 different varieties.

- Mendel noted that they differ from one another in many ways. Thus Mendel had chosen 7 pairs of contrasting characters for his study as shown in the table.

Characteristic studied	Dominant character	Recessive character
Stem length	Long	Short
Flower Position	Axillary	Terminal
Flower colour	Blue	White
Pod shape	Inflated	Constricted
Pod colour	Green	Yellow
Seed shape	Round	Wrinkled
Seed colour	Yellow	Green

Constrasting characters of pea plant used by Mendel

Reasons for Mendel's success

He chose the pea plant as it was advantageous for experimental work in many aspects

1. It is naturally self-pollinating and so is very easy to raise pure breeding individuals.

2. It has a short life span as it is an annual and so it was possible to follow several generations.

3. It is easy to cross-pollinate.

4. It has deeply defined contrasting characters.

5. The flowers are bisexual.

Monohybrid Cross – Inheritance of One Gene

Crosses involving **inheritance of only one pair of contrasting characters** are called monohybrid crosses. For example it is a cross between two forms of a single trait like a cross between tall and dwarf plant.

Mendel's Explanation of Monohybrid Cross Parental generation:

- Pure breeding tall plant and a pure breeding dwarf plant.

F1 generation:

- Plants raised from the seeds of pure breeding parental cross in F1 generation were tall and monohybrids.

F2 generation:

- Selfing of the F1 monohybrids resulted in tall and dwarf plants respectively in the ratio of 3:1. The actual number of tall and dwarf plants obtained by Mendel was 787 tall and 277 dwarf.

- External expression of a particular trait is known as phenotype. So the phenotypic ratio is 3:1.

- In the F2 generation 3 different types were obtained:
  Tall Homozygous – TT (Pure) – 1
  Tall Heterozygous – Tt – 2
  Dwarf Homozygous – tt – 1

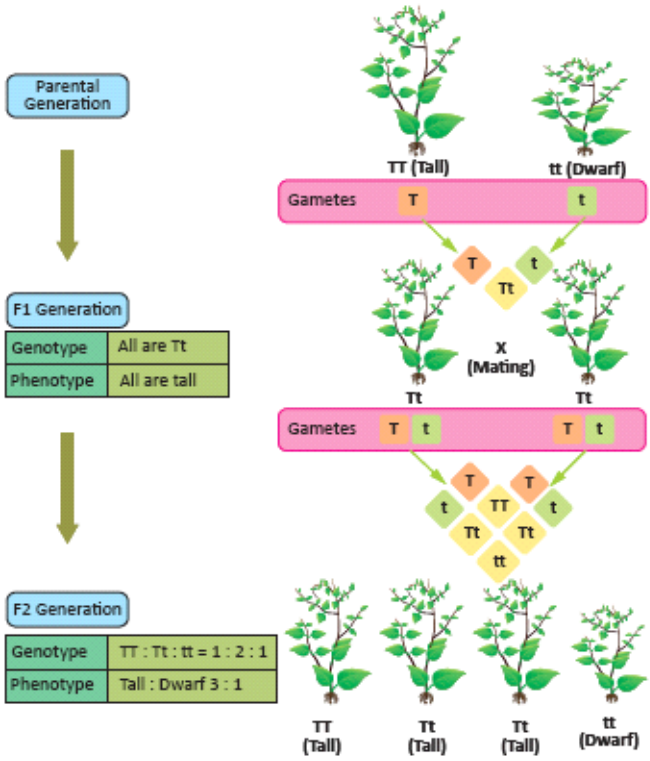
- So the **genotypic ratio 1:2:1**. A genotype is the genetic expression of an organism

Mendel's Interpretation on Monohybrid cross

- Based on these observations it was confirmed by Mendel that 'factors' are passed on from one generation to another, **now refered to as genes**.

- Tallness and Dwarfness are determined by a pair of contrasting factors , tall plant possess a pair of factors (represented by T- taking the first letter of the dominant character) and a plant is dwarf because it possess factors for dwarfness (represented as t- recessive character).

- These factors occur in pairs and may be alike as in pure breeding tall plants (TT) and dwarf plants (tt). This is referred to as **homozygous**. If they are unlike (Tt) they are referred to as **heterozygous**.



Monohybrid cross

1. Two factors making up a pair of contrasting characters are called **alleles.** Phenotypic expression of alleles are called **allelomorphs**. One member of each pair is contributed by one parent.

2. When two factors for alternative expression of a trait are brought together by fertilization. The **character which expresses itself** is called **dominant (Tallness)** condition and that which is **masked** is called **recessive condition (Dwarfness)**.

3. The factors are always pure and when gametes are formed, the unit factors segregate so that each gamete gets one of the two alternative factors. It means that factors for tallness (T) and dwarfness (t) are separate entities and in a gamete either T or t is present. When F1 hybrids are self crossed the two entities separate and then unite independently, forming tall and dwarf plants.

Dihybrid Cross- Inheritance Two Genes and Law of Independent Assortment

- Dihybrid cross involves the **inheritance of two pairs of contrasting characteristics** (or contrasting traits) at the same time.
- The two pairs of contrasting characteristics chosen by Mendel were **shape and color of seeds: round-yellow seeds and wrinkled-green seeds**.
- Mendel crossed pea plants having round yellow seeds with pea plants having wrinkled green seeds. Mendel made the following observations:

1. Mendel first crossed pure breeding pea plants having round-yellow seeds with pure breeding pea plants having wrinkled green seeds and found that only round yellow seeds were produced in the first generation (F1). No wrinkled-green seeds were obtained in the F1 generation. From this it was concluded that **round shape and yellow color of the seeds were dominant traits over the wrinkled shape and green color of the seeds.**

2. When the hybrids of F1 generation pea plants having round-yellow seeds were cross-bred by self pollination, then four types of seeds having different combinations of shape and color were obtained in second generation or F2 generation. They were **round yellow**, **round-green**, **wrinkled yellow** and wrinkled-green seeds.

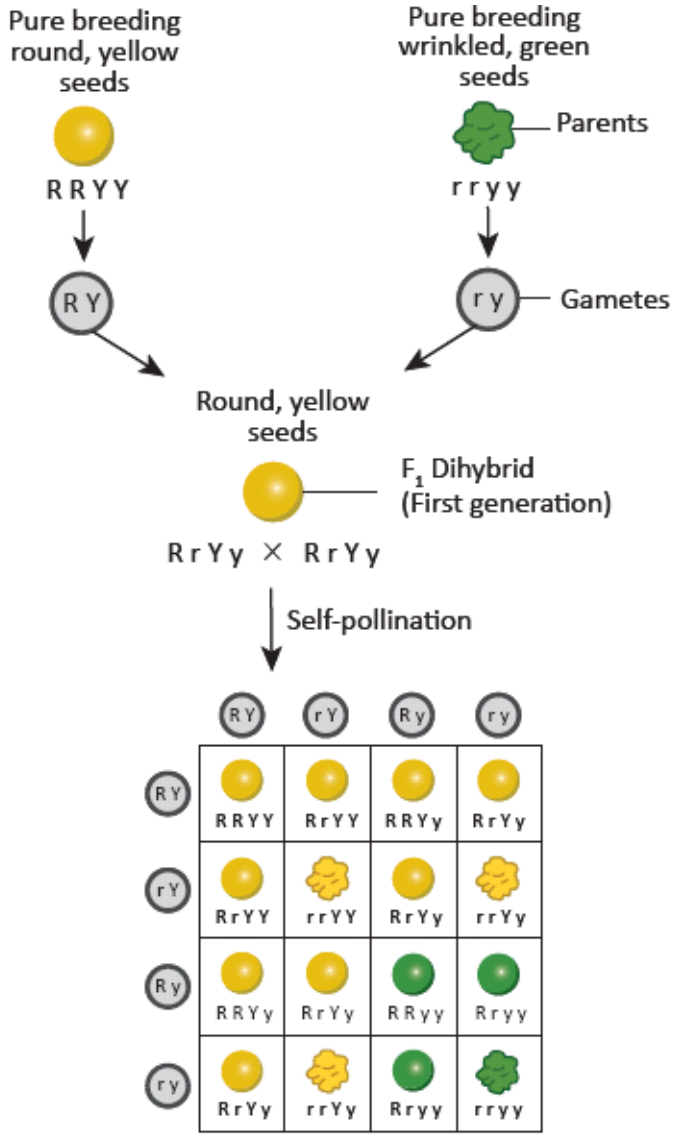
- The ratio of each phenotype (or appearance) of seeds in the **F2 generation is 9:3:3:1**. This is known as the **Dihybrid ratio**.

- From the above results it can be concluded that the factors for each character or trait remain independent and maintain their identity in the gametes. The factors are independent to each other and pass to the offsprings (through gametes).

Results of a Dihybrid Cross:
Mendel got the following results from his dihybrid cross

1. Four Types of Plants: A dihybrid cross produced four types of F2 offsprings in the ratio of 9 with two dominant traits, 3 with one dominant trait and one recessive trait, 3 with another dominant trait and another recessive trait and 1 with two recessive traits.

2. New Combination: Two new combinations of traits with round green and wrinkled yellow had appeared in the dihybrid cross (F2 generation).



Phenotypic ratio of F2 generation - 9:3:3:1

Round, Yellow – 9 Round, Green - 3
Wrinkled, Yellow – 3 Wrinkled, Green – 1

Dihybrid Cross

Mendel's Laws

Based on his experiments of monohybrid and dihybrid cross, Mendel proposed three important laws which are now called as Mendel's **Laws of Heredity**.

Law of Dominance:

"When two homozygous individuals with one or more sets of contrasting characters are crossed, the characters that appear in the F1 hybrid are dominant and those that do not appear in F1 are recessive characters".

Law of Segregation or Law of purity of gametes:

"When a pair of contrasting factors are brought together in a F1 hybrid. The two factors of the allelic pair remain together without mixing and when gametes are formed, the two separate out, so that only one enters each gamete."

Law of independent assortment:

"In case of inheritance of two or more pairs of characters simultaneously, the factors or genes of one pair assort out independently of the other pair."

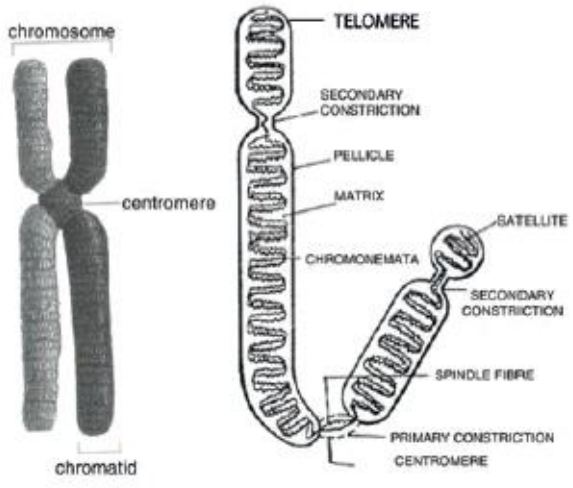
Chromosomes, DNA and Genes

- The human body is made up of million cells. The nucleus of each cell contains thin thread like structures called **chromosomes**.
- The term 'chromosomes' was first coined by **Waldeyer** in 1888. The chromosomes are the carrier of genetic material which contains the heredity information.
- The chromosomes are highly condensed coiled chromatin fibres packed with the **DNA** (Deoxyribonucleic acid) that forms the genetic material.
- **Genes** are segments of DNA, which are responsible for the inheritance of a particular phenotypic character.
- Each gene is present at a **specific position** on a chromosome called its **locus**. During cell division, the genetic information present in the genes are passed from one generation to another.

Structure of a Chromosome

- The chromosomes are thin, long and thread like structures consisting of two identical strands called sister chromatids.
- They are held together by the centromere. Each **chromatid** is made up of spirally coiled thin structure called **chromonema.**
- The chromonema has number of bead-like structures along its length which are called **chromomeres.**

- The chromosomes are made up of DNA, RNA, chromosomal proteins (histones and non-histones) and certain metallic ions. These proteins provide structural support to the chromosome.



Structure of chromosome

- A chromosome consists of the following regions

Primary constriction:

- The two arms of a chromosome meet at a point called **primary constriction** or **centromere**. The centromere is the region where spindle fibres attach to the chromosomes during cell division.

Secondary constriction:

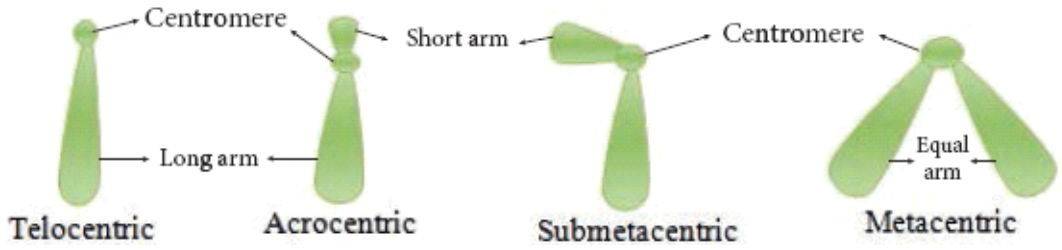
- Some chromosomes possess secondary constriction **at any point** of the chromosome. They are known as the nuclear zone or **nucleolar organizer** (formation of nucleolus in the nucleus).

Telomere:

- The **end of the chromosome** is called telomere. Each extremity of the chromosome has a polarity and prevents it from joining the adjacent chromosome. It maintains and provides **stability to the chromosomes.**

Satellite:

- Some of the chromosomes have an elongated **knob-like appendage** at one end of the chromosome known as satellite. The chromosomes with satellites are called as the **sat-chromosomes**.



Types of chromosomes based on position of centromere

Types of Chromosomes based on the position of Centromere

- Based on the position of centromere, the chromosomes are classified as **Telocentric, Acrocentric, Submetacentric** and **Metacentric**

1. Telocentric– The centromere is found on the proximal end. They are rod shaped chromosomes.

2. Acrocentric – The centromere is found at the one end with a short arm and a long arm. They are also rod-shaped chromosomes.

3. Submetacentric – The centromere is found near the centre of the chromosome. Thus forming two unequal arms. They are J shaped or L shaped chromosomes.

4. Metacentric – The centromere occurs in the centre of the chromosome and form two equal arms. They are V shaped chromosomes.

Types of Chromosomes based on function

- The eukaryotic chromosomes are classified into **autosomes** and **allosomes.**
- Autosomes contain genes that determine the somatic (body) characters**.** Male and female have equal number of autosomes**.**
- Allosomes are chromosomes which are responsible for determining the sex of an individual.
- They are also called as **sex chromosomes** or **hetero-chromosomes**. There are two types of sex chromosomes, X and Y- chromosomes. Human male have one X chromosome and one Y chromosome and human female have two X chromosomes.

Karyotype

- The number of chromosomes in any living organism (animal or plant) is constant. In human, each cell normally contains **23 pairs** of chromosomes.

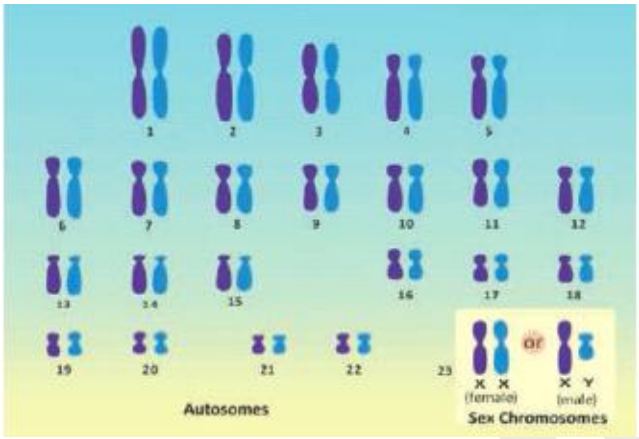
- Out of which 22 pairs are autosomes and the 23rd pair is the allosome or sex chromosome.

- In the body cells of sexually reproducing organisms, the chromosomes generally occur in pairs. This condition is called **diploid (2n)**.

- The gametes produced by the organisms contain a single set of chromosomes. Hence, the gametes are said to be **haploid (n).**

- Karyotype is the **number, size and shape of chromosomes** in the cell nucleus of an organism. **Idiogram** is the diagrammatic representation of karyotype of a species.

- It consists of all the metaphasic chromosomes arranged in homologous pairs according to decreasing length, thickness, position of centromere, shape etc., with the sex chromosomes placed at the end.



Normal human karyotype

Structure of DNA

- DNA is the hereditary material as it contains the genetic information. It is the most important constituent of a chromosome.

- The most widely accepted model of DNA is the double helical structure of **James Watson** and **Francis Crick**.

- They proposed the **three-dimensional model of DNA** on the basis of X-ray diffraction studies of DNA obtained by Rosalind Franklin and Maurice Wilkins.

- In appreciation of their discoveries on the molecular structure of nucleic acids Watson, Crick and Wilkins were awarded Nobel prize for Medicine in 1962.

Chemical Composition of DNA molecule

DNA is a large molecule consisting of millions of nucleotides. Hence, it is also called a **polynucleotide**. Each nucleotide consists of three components.

1. A sugar molecules – Deoxyribose sugar.
2. A nitrogenous base.

There are two types of nitrogenous bases in DNA. They are

(a) Purines (Adenine and Guanine)
(b) Pyrimidines (Cytosine and Thymine)

3. A phosphate group

Nucleoside and Nucleotide
Nucleoside = Nitrogen base + Sugar
Nucleotide = Nucleoside + Phosphate
The nucleotides are formed according to the purines and pyrimidines present in them.

Watson and Crick model of DNA

1. DNA molecule consists of two **polynucleotide** chains.

2. These chains form a **double helix** structure with two strands which run **anti-parallel** to one another.

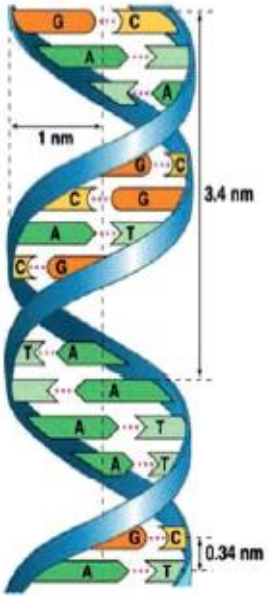
3. Nitrogenous bases in the centre are linked to **sugar-phosphate** units which form the backbone of the DNA.

4. Pairing between the nitrogenous bases is very specific and is always between purine and pyrimidine linked by hydrogen bonds.

Adenine (A) links Thymine (T) with two hydrogen bonds (A = T)

Cytosine (C) links Guanine (G) with three hydrogen bonds(C ≡ G)

This is called **complementary base pairing**.

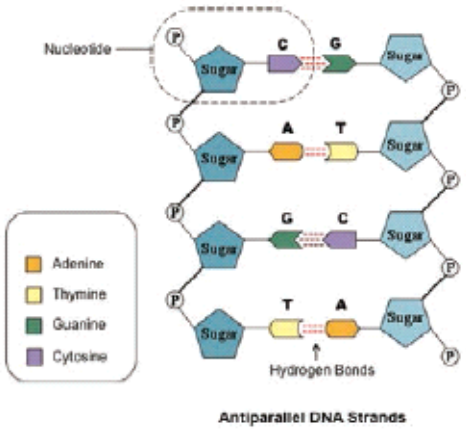


Structure of DNA

5. Hydrogen bonds between the nitrogenous bases make the DNA molecule stable.

6. Each turn of the double helix is 34 A° (3.4 nm). There are ten base pairs in a complete turn.

7. The nucleotides in a helix are joined together by phosphodiester bonds.



Nucleotides in a DNA

DNA Replication

- DNA replication is one of the basic process that occurs within a cell. DNA molecule produces exact copies of its own structure during replication process.
- The two strands of a DNA molecule have complementary base pairs, the nucleotides of each strand provide the information needed to produce its new strand.
- The two resulting daughter cells contain exactly the same genetic information as the parent cell. DNA replication involves the following steps

Origin of replication

- The specific point on the DNA, where the replication begins, is the **site of origin** of replication. The two strands open and separate at this point forming the **replication fork**.

Unwinding of DNA molecule

- The enzyme called **helicase**, bind to the origin of replication site. Helicase separates the two strands of the DNA.
- The enzyme called **topoisomerase** separates the double helix above the replication fork and removes the twists formed during the unwinding process. Each of the separated DNA strands function as a template.

Formation of RNA primer

- An RNA primer is a short segment of RNA nucleotides. The primer is synthesized by the DNA template close to the origin of replication site.

Synthesis of new complementary strand from the parent strand

- After the formation of RNA primer, nucleotides are added with the help of an enzyme **DNA polymerase** and a new complementary strand of DNA is formed from each of the parent strand. The synthesis is unidirectional.

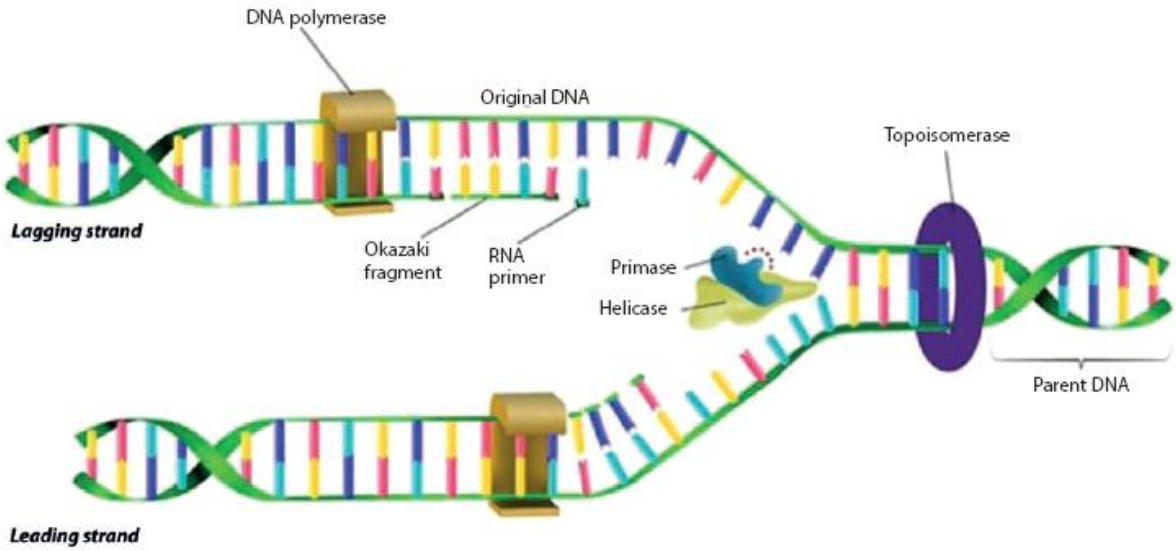
- In one strand, the daughter strand is synthesized as a continuous strand which is called **leading strand.**

- In the other strand, short segments of DNA are synthesized. This strand is called **lagging strand.** The short segments of DNA are called **Okazaki fragments**.
- The fragments are joined together by the enzyme, **DNA ligase**.

- The replication stops when the replication fork of the two sides meet at a site called **terminus**, which is situated opposite to origin of replication site.

Significance of DNA

- It is responsible for the transmission of hereditary information from one generation to next generation.



Replication of DNA

- It contains information required for the formation of proteins.

- It controls the developmental process and life activities of an organism.

Sex Determination

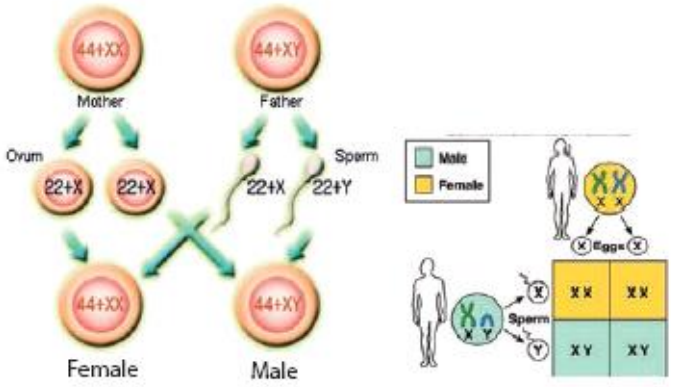
- The formation of zygote into male or female sex during development is called sex determination. Sex is determined by the chromosomes of an individual.

Sex Determination in Human

- Recall that human beings have 23 pairs of chromosomes out of which 22 pairs are autosomes and one pair (23rd pair) is the sex chromosome.

- The female gametes or the eggs formed are similar in their chromosome type (22+XX). Therefore, human females are **homogametic**.
- The male gametes or sperms produced are of two types. They are produced in equal proportions.

- The sperm bearing **(22+X)** chromosomes and the sperm bearing **(22+Y)** chromosomes. The human males are called **heterogametic.**



Sex determination in human

- It is a chance of probability as to which category of sperm fuses with the egg. If the egg (X) is fused by the X-bearing sperm an **XX individual (female)** is produced.

- If the egg (X) is fused by the Y-bearing sperm an **XY individual (male)** is produced. The sperm, produced by the father, determines the sex of the child. The mother is not responsible in determining the sex of the child.

- Now let's see how the chromosomes take part in this formation. Fertilization of the egg (22+X) with a sperm (22+X) will produce a female child (44+XX).

- While fertilization of the egg (22+X) with a sperm (22+Y) will give rise to a male child (44+XY).

Mutation

- The term mutation was introduced by **Hugo De Vries** in 1901 when he observed phenotypic changes in the evening primrose plant, Oenothera lamarckiana.

- Mutation is an inheritable sudden change in the genetic material (DNA) of an organism. Mutations are classified into two main types, namely chromosomal mutation and gene mutation.

1. **Chromosomal mutation**

- The **sudden change** in the **structure** or **number of chromosomes** is called chromosomal mutation. This may result in

(i) Changes in the structure of chromosomes:

- A structural change in the chromosomes usually occurs due to errors in cell division.

- Changes in the number and arrangement of genes takes place as a result of deletion, duplication, inversion and translocation in chromosomes.

(ii) Changes in the number of chromosomes:

They involve addition or deletion in the number of chromosomes present in a cell. This is called **ploidy**. There are two types of ploidy

(a) Euploidy
(b) Aneuploidy.

Euploidy:

- It is the condition in which the individual bears **more than the usual number** of diploid (2n) chromosomes. If an individual has three haploid sets of chromosomes, the condition is called **triploidy** (3n).

- Triploid plants and animals are typically sterile. If it has four haploid sets of chromosomes, the condition is called **tetraploidy** (4n).

- Tetraploid plants are advantageous as they often result in increased fruit and flower size.

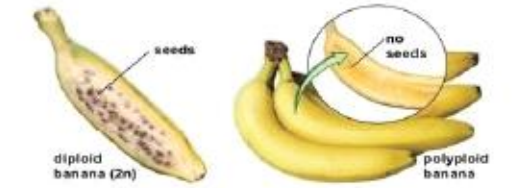
Aneuploidy:

- It is the **loss** or **gain** of **one** or **more chromosomes** in a set. It is of three types.

- **Monosomy** (2n 1), **Trisomy** (2n+1) and **Nullisomy** (2n-2). In man, Down's syndrome is one of the commonly known aneuploid conditions.

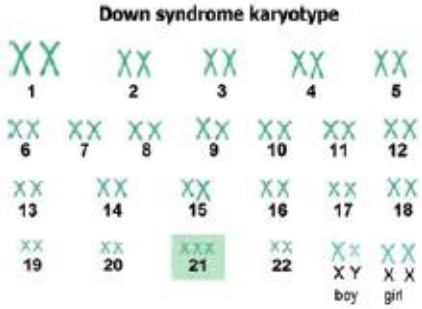
Down's syndrome

- This condition was first identified by a doctor named **Langdon Down** in 1866.
- It is a genetic condition in which there is an extra copy of **chromosome 21 (Trisomy 21)**. It is associated with mental retardation, delayed development, behavioural problems, weak muscle tone, vision and hearing disability are some of the conditions seen in these children.



Triploid Oyster Diploid Oyster

Euploidy



Aneuploidy

2. Gene or point mutation

- Gene mutation is the **changes** occurring in **nucleotide sequence of a gene**.
- It involves substitution, deletion, insertion or inversion of a single or more than one nitrogenous base.
- Gene alteration results in abnormal protein formation in an organism.

More to know:

- Punnett square is a checker board form devised by a British geneticist R.C.Punnett for study of genetics. It is a graphical representation to calculate the probability of all possible genotypes of offsprings in a genetic cross.

- T.H. Morgan was awarded Nobel Prize in 1993 for determining the role of chromosomes in heredity.

- **Telomeres act as aging clock in every cell.** Telomeres are protective sequences of nucleotides found in chromosomes. As a cell divides every time, they become shorter. Telomeres get too short to do their job, causing our cells to age.

- **Chargaff rule of DNA base pairing Erwin Chargaff** states that in DNA, the proportion of adenine is always equal to that of thymine and the proportion of guanine always equal to that of cytosine.

- S**ickle cell anaemia** is caused by the mutation of a single gene. Alteration in the gene brings a change in the structure of the protein part of haemoglobin molecule. Due to the change in the protein molecule, the red blood cell (RBC) that carries the haemoglobin is sickle shaped.